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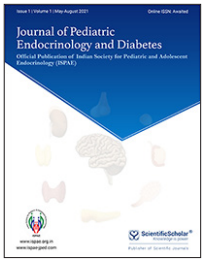

Clinical Images

A short child with white bones

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Quick Response Code:

A 10-year-old boy presented with poor weight and height gain since early childhood without prior systemic illness or clinical features suggesting a specific etiology. He was born at full term through vaginal delivery with a birth weight of 2.5 kg and the perinatal and neonatal periods were unremarkable. The patient had a single right tibial fracture following a trivial trauma during early childhood.

Examination revealed frontal bossing, prominent eyes with blue sclerae, dental crowding [Figure 1a], brachydactyly with soft finger pads, and spoon-shaped nails [Figure 1b]. Anthropometric assessment showed proportionate short stature (upper segment to lower segment ratio of 1:1 and height Z score −2.61). The skeletal survey showed acro-osteolysis [Figure 1c], open posterior fontanelle [Figure 1d], osteosclerosis [Figure 1e], and notching of vertebrae [Figure 1f]. Growth hormone (GH) stimulation testing confirmed normal GH secretion. The clinical and imaging findings were consistent with pycnodysostosis (PYCD), a rare skeletal dysplasia. Genetics was planned, but could not be done due to financial constraints.

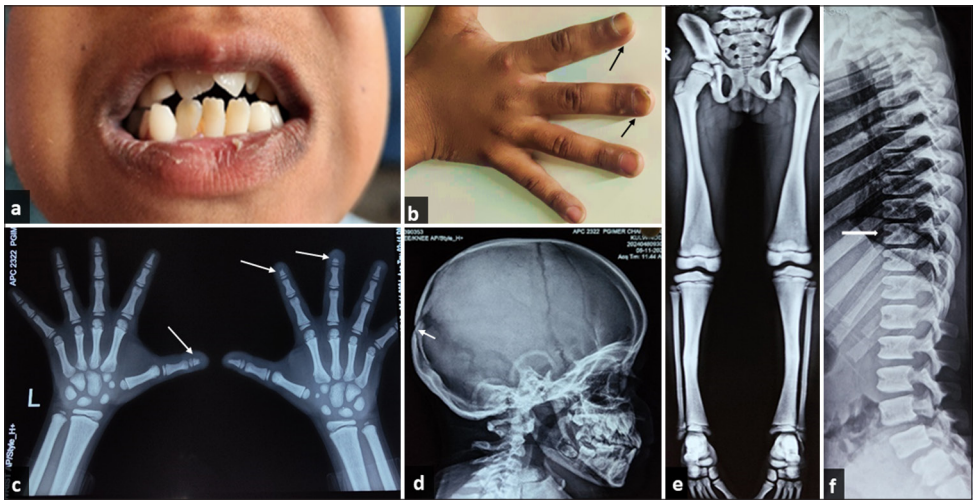

Figure 1: A 10-year-old boy with pycnodysostosis. (a) Dental crowding, (b) spoon-shaped nails (black arrows), (c) acro-osteolysis (white arrows), (d) open posterior fontanelle (white arrow), (e) osteosclerosis, and (f) notching of vertebrae (white arrow).

PYCD is a rare autosomal recessive skeletal dysplasia caused by pathogenic cathepsin K (*CTSK*) gene variants, resulting in defective CTSK activity.[1] Diagnosis relies on clinical features and pathognomonic radiographic findings of osteosclerosis with acro-osteolysis. Common morbidities include fracture risk, dental abnormalities, and growth failure.[1,2] The differential diagnoses for PYCD include osteopetrosis, cleidocranial dysplasia, and idiopathic acro-osteolysis. Growth hormone stimulation testing is advised for severely stunted children as early treatment is advantageous.[2] Early diagnosis is vital for tailored management, including fracture prevention, dental care, and growth monitoring.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

Dr. Jaivinder Yadav is on the Editorial Board of the Journal.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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